



GAA gene

glucosidase alpha, acid

Normal Function

The *GAA* gene provides instructions for producing an enzyme called acid alpha-glucosidase (also known as acid maltase). This enzyme is active in lysosomes, which are structures that serve as recycling centers within cells. Lysosomes use digestive enzymes to break down complex molecules into simpler ones that can be used by cells. Acid alpha-glucosidase normally breaks down a complex sugar called glycogen into a simpler sugar called glucose. Glucose is the main energy source for most cells.

Health Conditions Related to Genetic Changes

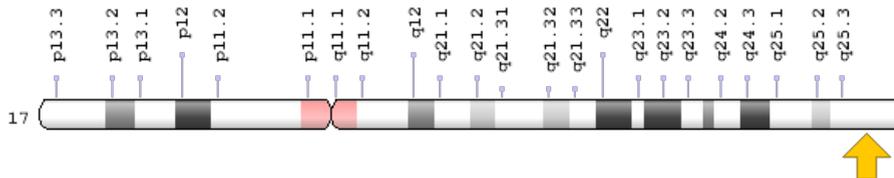
Pompe disease

More than 200 mutations in the *GAA* gene have been identified in people with Pompe disease. Many of these mutations change one of the protein building blocks (amino acids) used to make acid alpha-glucosidase. Other mutations insert or delete genetic material in the *GAA* gene. Mutations in this gene significantly reduce the activity of acid alpha-glucosidase, preventing the enzyme from breaking down glycogen effectively. As a result, this complex sugar can build up to toxic levels in lysosomes. The abnormal buildup of glycogen damages organs and tissues throughout the body, particularly the muscles, leading to progressive muscle weakness, heart problems, and the other features of Pompe disease.

Chromosomal Location

Cytogenetic Location: 17q25.3, which is the long (q) arm of chromosome 17 at position 25.3

Molecular Location: base pairs 80,101,526 to 80,119,882 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- acid alpha-glucosidase
- acid alpha-glucosidase preproprotein
- acid maltase
- Aglucosidase alfa
- Alpha-1,4-glucosidase
- Amyloglucosidase
- Glucoamylase
- glucosidase, alpha; acid
- glucosidase, alpha; acid (Pompe disease, glycogen storage disease type II)
- LYAG
- LYAG_HUMAN
- lysosomal alpha-glucosidase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Glycogen-Engorged Lysosome (image)
<https://www.ncbi.nlm.nih.gov/books/NBK22444/?rendertype=figure&id=A2966>
- The Cell: A Molecular Approach (second edition, 2000): Lysosomes
<https://www.ncbi.nlm.nih.gov/books/NBK9953/>

GeneReviews

- Glycogen Storage Disease Type II (Pompe Disease)
<https://www.ncbi.nlm.nih.gov/books/NBK1261>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%22GAA+gene%22%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- GLUCOSIDASE, ALPHA, ACID
<http://omim.org/entry/606800>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=GAA%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4065
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2548>
- UniProt
<http://www.uniprot.org/uniprot/P10253>

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